SEQUENCE LISTING

<110> Assistance Publique - Hopitaux de Paris (AH-HP)
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      Universite de Versailles - Saint-Quentin-en-Yvelines
      Universite Paris-Sud
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      UGO, Valerie
      JAMES, Chloe
      LE COUEDIC, Jean-Pierre
      CASADEVALL, Nicole

<120> Identification of a JAK2 mutation involved in Vaquez
      Polyglobulia

<130> D 22707

<140> FR04-11480
<141> 2004-10-27

<160> 31

<170> PatentIn version 3.3

<210> 1
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<212> PRT
<213> homo sapiens

<220>
<223> variant JAK2 V617F

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Lys Gln Ile Asp Pro Val Leu Gln Val Tyr Leu Tyr His Ser Leu Gly
            35   40   45

Lys Ser Glu Ala Asp Tyr Leu Thr Phe Pro Ser Gly Glu Tyr Val Ala
        50   55   60

Glu Glu Ile Cys Ile Ala Ala Ser Lys Ala Cys Gly Ile Thr Pro Val
65   70   75   80

Tyr His Asn Met Phe Ala Leu Met Ser Glu Thr Glu Arg Ile Trp Tyr
                85   90   95

Pro Pro Asn His Val Phe His Ile Asp Glu Ser Thr Arg His Asn Val
            100   105   110

Leu Tyr Arg Ile Arg Phe Tyr Phe Pro Arg Trp Tyr Cys Ser Gly Ser
            115   120   125

Asn Arg Ala Tyr Arg His Gly Ile Ser Arg Gly Ala Glu Ala Pro Leu
    130   135   140

Leu Asp Asp Phe Val Met Ser Tyr Leu Phe Ala Gln Trp Arg His Asp
145   150   155   160

Phe Val His Gly Trp Ile Lys Val Pro Val Thr His Glu Thr Gln Glu
            165   170   175

Glu Cys Leu Gly Met Ala Val Leu Asp Met Met Arg Ile Ala Lys Glu
        180   185   190

Asn Asp Gln Thr Pro Leu Ala Ile Tyr Asn Ser Ile Ser Tyr Lys Thr
        195   200   205

Phe Leu Pro Lys Cys Ile Arg Ala Lys Ile Gln Asp Tyr His Ile Leu
    210   215   220

Thr Arg Lys Arg Ile Arg Tyr Arg Phe Arg Arg Phe Ile Gln Gln Phe
225   230   235   240

Ser Gln Cys Lys Ala Thr Ala Arg Asn Leu Lys Leu Lys Tyr Leu Ile
            245   250   255

Asn Leu Glu Thr Leu Gln Ser Ala Phe Tyr Thr Glu Lys Phe Glu Val
        260   265   270

Lys Glu Pro Gly Ser Gly Pro Ser Gly Glu Glu Ile Phe Ala Thr Ile
        275   280   285

Ile Ile Thr Gly Asn Gly Gly Ile Gln Trp Ser Arg Gly Lys His Lys
    290   295   300

Glu Ser Glu Thr Leu Thr Glu Gln Asp Leu Gln Leu Tyr Cys Asp Phe
305   310   315   320

Pro Asn Ile Ile Asp Val Ser Ile Lys Gln Ala Asn Gln Glu Gly Ser
            325   330   335

Asn Glu Ser Arg Val Val Thr Ile His Lys Gln Asp Gly Lys Asn Leu
        340   345   350

Glu Ile Glu Leu Ser Ser Leu Arg Glu Ala Leu Ser Phe Val Ser Leu
        355   360   365

Ile Asp Gly Tyr Tyr Arg Leu Thr Ala Asp Ala His His Tyr Leu Cys
    370   375   380

Lys Glu Val Ala Pro Pro Ala Val Leu Glu Asn Ile Gln Ser Asn Cys
385   390   395   400

His Gly Pro Ile Ser Met Asp Phe Ala Ile Ser Lys Leu Lys Lys Ala
            405   410   415

Gly Asn Gln Thr Gly Leu Tyr Val Leu Arg Cys Ser Pro Lys Asp Phe
        420   425   430

Asn Lys Tyr Phe Leu Thr Phe Ala Val Glu Arg Glu Asn Val Ile Glu
        435   440   445

Tyr Lys His Cys Leu Ile Thr Lys Asn Glu Asn Glu Glu Tyr Asn Leu

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Ser Gly Thr Lys Lys Asn Phe Ser Ser Leu Lys Asp Leu Leu Asn Cys
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Tyr Gln Met Glu Thr Val Arg Ser Asp Asn Ile Ile Phe Gln Phe Thr
                485             490                 495

Lys Cys Cys Pro Pro Lys Pro Lys Asp Lys Ser Asn Leu Leu Val Phe
            500             505                 510

Arg Thr Asn Gly Val Ser Asp Val Pro Thr Ser Pro Thr Leu Gln Arg
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Pro Thr His Met Asn Gln Met Val Phe His Lys Ile Arg Asn Glu Asp
    530             535                 540

Leu Ile Phe Asn Glu Ser Leu Gly Gln Gly Thr Phe Thr Lys Ile Phe
545             550                 555                 560

Lys Gly Val Arg Arg Glu Val Gly Asp Tyr Gly Gln Leu His Glu Thr
            565             570                 575

Glu Val Leu Leu Lys Val Leu Asp Lys Ala His Arg Asn Tyr Ser Glu
        580             585                 590

Ser Phe Phe Glu Ala Ala Ser Met Met Ser Lys Leu Ser His Lys His
        595         .       600                 605

Leu Val Leu Asn Tyr Gly Val Cys Phe Cys Gly Asp Glu Asn Ile Leu
    610             615                 620

Val Gln Glu Phe Val Lys Phe Gly Ser Leu Asp Thr Tyr Leu Lys Lys
625             630                 635                 640

Asn Lys Asn Cys Ile Asn Ile Leu Trp Lys Leu Glu Val Ala Lys Gln
            645             650                 655

Leu Ala Trp Ala Met His Phe Leu Glu Glu Asn Thr Leu Ile His Gly
        660             665                 670

Asn Val Cys Ala Lys Asn Ile Leu Leu Ile Arg Glu Glu Asp Arg Lys
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Thr Gly Asn Pro Pro Phe Ile Lys Leu Ser Asp Pro Gly Ile Ser Ile
    690             695                 700

Thr Val Leu Pro Lys Asp Ile Leu Gln Glu Arg Ile Pro Trp Val Pro
705             710                 715                 720

Pro Glu Cys Ile Glu Asn Pro Lys Asn Leu Asn Leu Ala Thr Asp Lys
            725             730                 735

Trp Ser Phe Gly Thr Thr Leu Trp Glu Ile Cys Ser Gly Gly Asp Lys
            740             745                 750

Pro Leu Ser Ala Leu Asp Ser Gln Arg Lys Leu Gln Phe Tyr Glu Asp
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Arg His Gln Leu Pro Ala Pro Lys Trp Ala Glu Leu Ala Asn Leu Ile
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Ile Ile Arg Asp Leu Asn Ser Leu Phe Thr Pro Asp Tyr Glu Leu Leu
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Thr Glu Asn Asp Met Leu Pro Asn Met Arg Ile Gly Ala Leu Gly Phe
            820   825   830

Ser Gly Ala Phe Glu Asp Arg Asp Pro Thr Gln Phe Glu Glu Arg His
        835   840   845

Leu Lys Phe Leu Gln Gln Leu Gly Lys Gly Asn Phe Gly Ser Val Glu
    850   855   860

Met Cys Arg Tyr Asp Pro Leu Gln Asp Asn Thr Gly Glu Val Val Ala
865   870   875   880

Val Lys Lys Leu Gln His Ser Thr Glu Glu His Leu Arg Asp Phe Glu
            885   890   895

Arg Glu Ile Glu Ile Leu Lys Ser Leu Gln His Asp Asn Ile Val Lys
        900   905   910

Tyr Lys Gly Val Cys Tyr Ser Ala Gly Arg Arg Asn Leu Lys Leu Ile
    915   920   925

Met Glu Tyr Leu Pro Tyr Gly Ser Leu Arg Asp Tyr Leu Gln Lys His
    930   935   940

Lys Glu Arg Ile Asp His Ile Lys Leu Leu Gln Tyr Thr Ser Gln Ile
945   950   955   960

Cys Lys Gly Met Glu Tyr Leu Gly Thr Lys Arg Tyr Ile His Arg Asp
            965   970   975

Leu Ala Thr Arg Asn Ile Leu Val Glu Asn Glu Asn Arg Val Lys Ile
        980   985   990

Gly Asp Phe Gly Leu Thr Lys Val Leu Pro Gln Asp Lys Glu Tyr Tyr
    995   1000   1005

Lys Val Lys Glu Pro Gly Glu Ser Pro Ile Phe Trp Tyr Ala Pro
    1010   1015   1020

Glu Ser Leu Thr Glu Ser Lys Phe Ser Val Ala Ser Asp Val Trp
    1025   1030   1035

Ser Phe Gly Val Val Leu Tyr Glu Leu Phe Thr Tyr Ile Glu Lys
    1040   1045   1050

Ser Lys Ser Pro Pro Ala Glu Phe Met Arg Met Ile Gly Asn Asp
    1055   1060   1065

Lys Gln Gly Gln Met Ile Val Phe His Leu Ile Glu Leu Leu Lys
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Asn Asn Gly Arg Leu Pro Arg Pro Asp Gly Cys Pro Asp Glu Ile
    1085   1090   1095

Tyr Met Ile Met Thr Glu Cys Trp Asn Asn Asn Val Asn Gln Arg
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Pro Ser Phe Arg Asp Leu Ala Leu Arg Val Asp Gln Ile Arg Asp
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Asn Met Ala Gly
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<223> G1849T mutation in jak2 gene

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gctgctgcgg cgcagggaga ggcctggtcc tcgctgccga gggatgtgag tgggagctga   240

gcccacactg gagggccccc gagggcccag cctggaggtc gttcagagcc gtgcccgccc   300

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gaaaaagact ctgcatggga atggcctgcc ttacgatgac agaaatggag ggaacatcca   540

cctcttctat atatcagaat ggtgatattt ctggaaatgc caattctatg aagcaaatag   600

atccagttct tcaggtgtat ctttaccatt cccttgggaa atctgaggca gattatctga   660

cctttccatc tggggagtat gttgcagaag aaatctgtat tgctgcttct aaagcttgtg   720

gtatcacacc tgtgtatcat aatatgtttg ctttaatgag tgaaacagaa aggatctggt   780

atccacccaa ccatgtcttc catatagatg agtcaaccag gcataatgta ctctacagaa   840

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tatctcgagg tgctgaagct cctcttcttg atgactttgt catgtcttac ctctttgctc   960

agtggcggca tgattttgtg cacggatgga taaaagtacc tgtgactcat gaaacacagg   1020

aagaatgtct tgggatggca gtgttagata tgatgagaat agccaaagaa aacgatcaaa   1080

ccccactggc catctataac tctatcagct acaagacatt cttaccaaaa tgtattcgag   1140

caaagatcca agactatcat attttgacaa ggaagcgaat aaggtacaga tttcgcagat   1200

ttattcagca attcagccaa tgcaaagcca ctgccagaaa cttgaaactt aagtatctta   1260

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tatattgcga ttttcctaat attattgatg tcagtattaa gcaagcaaac caagagggtt   1500

caaatgaaag ccgagttgta actatccata agcaagatgg taaaaatctg gaaattgaac   1560

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tacaaagcaa ctgtcatggc ccaatttcga tggattttgc cattagtaaa ctgaagaaag   1740

caggtaatca gactggactg tatgtacttc gatgcagtcc taaggacttt aataaatatt   1800

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atcttttgaa ttgttaccag atggaaactg ttcgctcaga caatataatt ttccagttta   1980

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gacctgaaaa aattattatg taaattttgc aatgttaaag atgcacagaa tatgtatgta    4260

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acttcactat acaaacaaat taagatgttc agataattga ataagtacct ttgtgtcctt    4440

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tttattatgg tttcccttgt atctatttgt ggtgaatgtg ttttttaaat ggaactatct    4680

ccaaattttt ctaagactac tatgaacagt tttctttttaa aattttgaga ttaagaatgc    4740

caggaatatt gtcatccttt gagctgctga ctgccaataa cattcttcga tctctgggat    4800

ttatgctcat gaactaaatt taagcttaag ccataaaata gattagattg ttttttaaaa    4860

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attcagagtc tttctttgaa gcagcaagta tgatgagcaa gctttctcac aagcatttgg     240

ttttaaatta tggagtatgt ttctgtggag acgagaatat tctggttcag gagtttgtaa     300

aatttggatc actagataca tatctgaaaa agaataaaaa ttgtataaat atattatgga     360

aacttgaagt tgctaaacag ttggcatggg ccatgcattt tctagaagaa aacaccctta     420

ttcatgggaa tgtatgtgcc aaaaatattc tgcttatcag agaagaagac aggaagacag     480

gaaatcctcc tttcatcaaa cttagtgatc ctggcattag tattacagtt ttgccaaagg     540

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cgagaatatt ctggttcagg agtttgtaaa attt                                  94
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<210> 6
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<223> PCR primer (55240-55260)

<400> 6
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<210> 7
<211> 20
<212> DNA
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<220>
<223> sequencing primer (54813-54832)

<400> 7
cagaacgttg atggcagttg                                             20


<210> 8
<211> 27
<212> DNA
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<220>
<223> sequencing primer (55207-55233)

<400> 8
tgaatagtcc tacagtgttt tcagttt                                     27


<210> 9
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<220>
<223> PCR and sequencing primer (1386-1407)

<400> 9
caacctcagt gggacaaaga a                                           21


<210> 10
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<220>
<223> PCR and sequencing primer (2019-2041)

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gcagaatatt tttggcacat aca   23

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<211> 42
<212> DNA
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<223> SNPS probes and detection of mutation and siRNA (1829-1870)

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<210> 12
<211> 23
<212> DNA
<213> homo sapiens

<220>
<223> sequence comprising the G1849T mutation

<400> 12
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<210> 13
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<220>
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<223> DNA portion of combined DNA/RNA sequence

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<223> n is T

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<223> DNA portion of combined DNA/RNA sequence

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<223> oligo "R" (antisens)

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<223> "Sensor wt"

<400> 17
gtctccacag acacatactc cataa   25

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<220>
<223> JAK2 primer

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<212> DNA
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<223> sequence of antisens primer

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<210> 31
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